t(9;12)(q22;p12)

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**Clinics and pathology**

**Disease**
Myelodysplastic syndrome

**Epidemiology**
Only one case to date

**Clinics**
A 36 yr old female patient

**Cytology**
Eosinophilia.

**Evolution**
Progressed to overt leukemia with megakaryocytic blasts.

**Cytogenetics**

**Additional anomalies**
Complex karyotype

**Genes involved and proteins**

**SYK**

**Location**
9q22

**Protein**
Contains SH2 domains and a protein kinase domain; non-receptor type protein-tyrosine kinase; role in signaling pathways.

**ETV6**

**Location**
12p13

**Protein**
Contains a Helix-Loop-Helix and ETS DNA binding domains; wide expression; nuclear localisation; ETS-related transcription factor.

**Result of the chromosomal anomaly**

**Hybrid gene**
Description
5' ETV6 - 3' SYK; the counterpart SYK-ETV6 is also expressed.

**Fusion protein**
Description
N term ETV6 exons 1 to 5 (with the Helix-Loop-Helix domain), fused to the C term SYK, containing the protein kinase domain.

**Oncogenesis**
ETV6-SYK is constitutively tyrosine phosphorylated.

**References**


This article should be referenced as such: